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EmbryoGenetics

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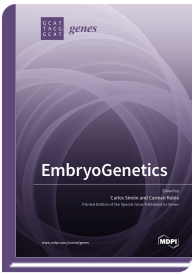

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The science of human genetics has advanced at an exponential pace since the double-helix structure of DNA was identified in 1953. Within only 25 years of that discovery, the first gene was sequenced. Subsequent efforts in the span of a few decades have brought advanced next-generation sequencing and new tools for genome editing, allowing scientists to write and rewrite the code of life. Importantly, with such rapid and sophisticated advancements, any tools or studies applicable to adult genetics can now also be applied to embryos. The first live births following preimplantation genetic testing (PGT) to identify sex in X-linked disease were reported by Alan Handyside in 1990. This groundbreaking work paved the way to extend the concept to PGT for monogenic diseases (PGT-M), including Mendelian single-gene defects, severe childhood lethality or early-onset disease, cancer predisposition, and HLA typing for histocompatible cord-blood stem cells' transplantation. Later, we moved onto the identification of aneuploidies in all 23 pairs of chromosomes, called PGT for aneuploidy (PGT-A). A step forward was the detection of chromosomal imbalances in carriers of structural chromosome rearrangements (PGT-SR). Another advancement came with PGT for polygenic risk scoring (PGT-P). Moreover, we are moving from embryo selection to intervention because the genetic code is not only readable, but also re-writeable. Indeed, gene editing is now possible using tools like CRISPR/Cas9, which are applicable to all species, including human embryos.

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